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Skills Practice Lab

Analyzing Karyotypes

OBJECTIVES

Make a human karyotype by arranging chromosomes in order by length, centromere position, and banding pattern.

Identify a karyotype as normal or abnormal.

Identify any genetic disorder that is present and describe the effect of the genetic disorder on the individual.

PROCESS SKILLS

- Identifying
- Describing
- Analyzing
- Modeling

MATERIALS

- chromosome spread
- human karyotyping form
- metric ruler
- photomicrograph of chromosomes
- scissors
- transparent tape

Background

1. Define and describe a human karyotype.

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2. Compare the two procedures used to obtain the cells needed for preparing a fetal karyotype.

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_____

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3. List four types of chromosomal mutations that can be identified by preparing a fetal karyotype.

_____

_____

4. Name the disorder that occurs in people who have an extra copy of chromosome 21.

_____

Procedure

1. Obtain a photomicrograph and note the letter identifying which person the cells were taken from.
2. **CAUTION Always cut in a direction away from your face and body.** Carefully cut apart the chromosomes on each photomicrograph. Be sure to leave a slight margin around each chromosome.
3. Arrange the chromosomes in homologous pairs. The members of each pair will be the same length and will have their centromeres located in the same area. Use the ruler to measure the length of the chromosome and the position of the centromere. The banding patterns of homologous chromosomes will be similar and may also help you pair the chromosomes.
4. Arrange the pairs according to their length. Begin with the largest chromosomes and move to the smallest.
5. Tape each pair of homologous chromosomes to a human karyotyping form. Place the centromeres on the lines provided. Place the longest chromosome at position 1, and the shortest at position 22. Place the two sex chromosomes at position 23.
6. The diagram you have made is a karyotype, as in **Figure 1**. Analyze your karyotype to determine the sex of the individual. Use the information in Table 1 to guide your analysis.

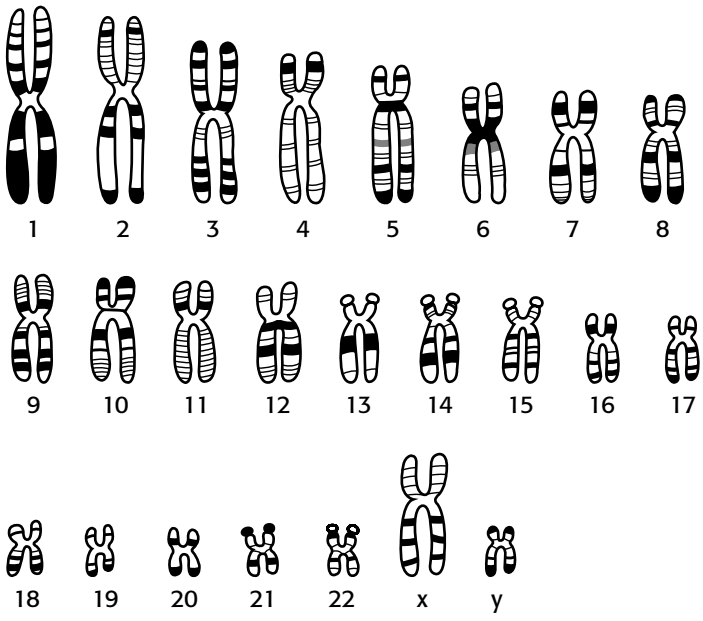

Figure 1 Normal Human Karyotype

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7. Record your results in Table 2. Pool your data with that from the rest of the class.

Table 1 Genetic Disorders Caused by an Abnormal Chromosome Number

Name of abnormality	Chromosome affected	Description of abnormality
Down syndrome, or Trisomy 21	#21	47 chromosomes; mental retardation with specific characteristic features; may have heart defects and respiratory problems
Edwards' syndrome, or Trisomy 18	#18	47 chromosomes; severe mental retardation; very characteristic malformations of the skull, pelvis, and feet, among others; die in early infancy
Patau syndrome, or Trisomy 13	#13	47 chromosomes; abnormal brain function that is very severe; many facial malformations; usually die in early infancy
Turner's syndrome	Single X in female (XO)	45 chromosomes; in females only; missing an X chromosome; do not develop secondary sex characteristics; are infertile
Klinefelter's syndrome	Extra X in male (XXY)	47 chromosomes; in males only; sterile, small testicles; otherwise normal appearance
XYY syndrome	Extra Y in male (XYY)	47 chromosomes; in males only; low mental ability; otherwise normal appearance
Triple X syndrome	Extra X in female (XXX)	47 chromosomes; sterility sometimes occurs; normal mental ability

Table 2 Pooled Class Data

Letter identifier	Sex	Condition	Chromosome abnormality
A			
B			
C			
D			
E			
F			
G			
H			
I			

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8. Clean up your materials and wash your hands before leaving the lab.

Analysis and Conclusions

1. Is the fetus represented by your karyotype male or female? How do you know?

2. Does the fetus have a genetic disorder? Explain.

3. Assume that two students started with the same photomicrograph. One student concluded that the individual had Down syndrome. The other student concluded that the individual had Edwards' syndrome. Explain how this could happen.

4. How is sex determined in a person who has more than two sex chromosomes? Explain your answer.

5. In this lab, you examined karyotypes for the presence of abnormal chromosome numbers in both autosomes and sex chromosomes. Which condition seems to have a greater influence on a person's health—trisomy of an autosome or trisomy of a sex chromosome?

6. Assume that an individual has a deletion mutation in one of their chromosomes. What would the karyotype look like in this situation?

7. How might banding patterns be important to detecting an inversion mutation?

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8. Some medical labs make karyotypes from several of an individual's cells before drawing conclusions about the individual's health. Do you think this is necessary? Why or why not?

Further Inquiry

Trisomy occurs when an individual has three copies of the same chromosome. Monosomy occurs when an individual has only one copy of a chromosome. In this lab, you examined a fetal karyotype for the presence of three different trisomies. Find out why monosomies are rarely detected.